

## 비전형적인 다발성 신경섬유종증 1예

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김현직 · 장정현 · 이현석 · 최홍식

### A Case of the Variant Type Neurofibromatosis

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#### ABSTRACT

A case of neurofibromatosis in a 51 year old woman, with no other evidences of genetic defect, is described. Neurofibromatosis affects primarily cell growth of neural tissues and can cause tumors to grow on nerves. In the past, neurofibromatosis has been divided into 2 groups, Type I and Type II. The two groups have been shown to be distinct at clinical and molecular levels. Cafe-au-lait spots, neurofibroma, freckling in axilla, lisch nodule, family history and defect of chromosome 17q are the distinctive features of neurofibromatosis Type I, whereas bilateral vestibular schwannoma, family history and defect of chromosome 22q characterize the neurofibromatosis Type II. We experienced a case of neurofibromatosis having multiple neurofibromas in both neck, left thoracic inlet, and spinal cord without abnormal chromosomal findings. We present this case with a review of the literature. (Korean J Otolaryngol 2004;47:1315-8)

KEY WORDS : Neurofibromatosis · Chromosome.

본 증례는 51세 여자 환자로, 유전적 결손 없이 다발성 신경섬유종증의 비전형적인 형태를 보였다. 신경섬유종증은 주로 신경 조직의 세포 성장을 촉진하여 신경에 종양이 자라게 한다. 과거에 신경섬유종증은 Type I과 Type II로 분류되어 왔으며, 임상적 및 분자적 수준에서 두 유형이 구별된다. Type I의 특징적인 특징으로는 cafe-au-lait 반점, 신경섬유종, 겨드랑이 색소침착, 리시 결절, 가족력 및 17q 염색체 결손이 포함된다. 반면 Type II의 특징적인 특징으로는 양측 전정신경종, 가족력 및 22q 염색체 결손이 포함된다. 본 증례는 목, 흉부 인접, 그리고 척수에서 다발성 신경섬유종이 관찰되었지만, 비정상적인 염색체 소견은 없었다. 본 증례를 문헌 고찰과 함께 보고한다. (Korean J Otolaryngol 2004;47:1315-8)

**키워드 :** Neurofibromatosis · Chromosome.

환자 1 (Von Recklinghausens disease) 17

cafe - au - lait (Fig. 1A) level 3 × 4 cm

(freckling), (iris harmatoma, Lisch spots) 2 22

(Fig. 1B)

3 4

(Fig. 2A, B and C) T2

high signal, T1 low signal, T1

가

(multiple schwannomatosis)

2003 5

20

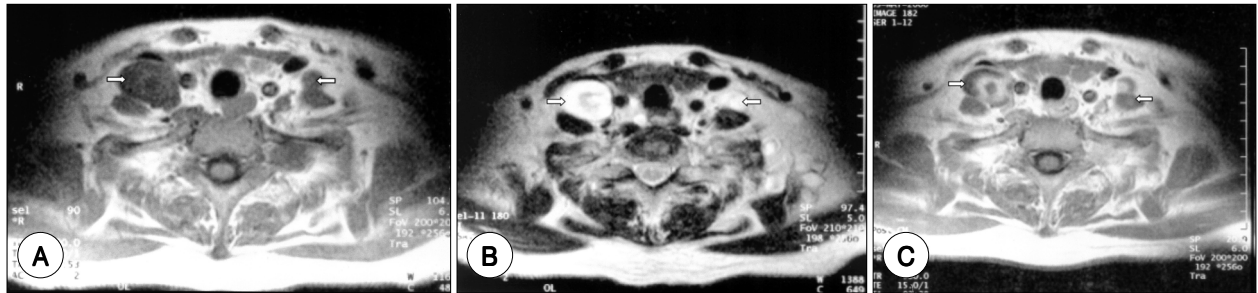
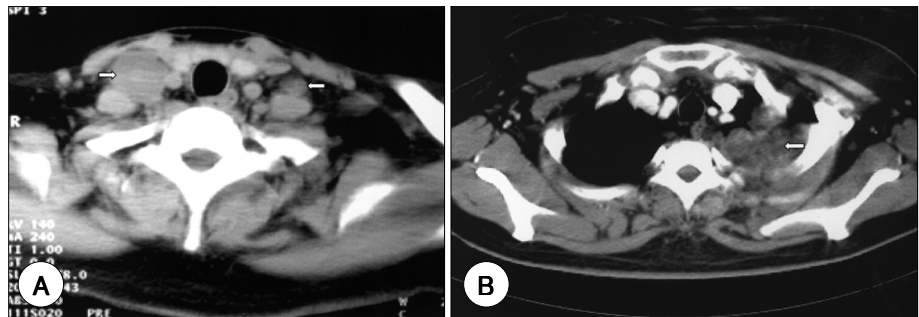
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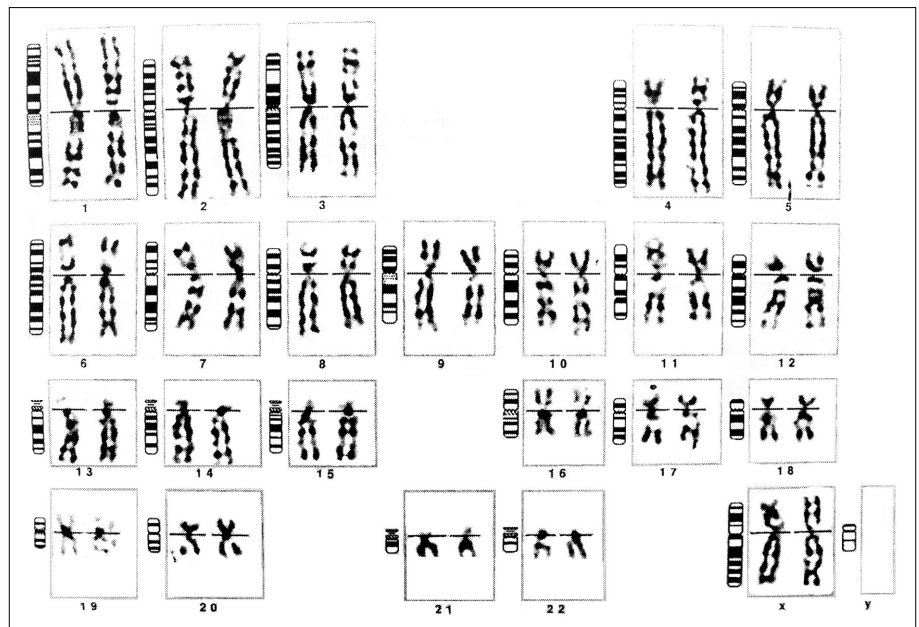
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**Fig. 1.** A : Neck CT shows 2.5×3 cm sized well defined homogenous mass lesion in right supraclavicular area and multiple variable sized mass lesion in both jugular chain. B : Chest CT shows multiple conglomerated central low density mass lesion in left lung apex, anterior and posterior chest wall.



**Fig. 2.** A : Neck MRI shows 2.5×3 cm sized well defined mass lesion in right supraclavicular area and multiple variable sized mass lesion in both jugular chain. It shows low signal in T1W image. B : Neck MRI shows high signal mass lesion in T2W image. C : Neck MRI shows well enhanced mass lesion in T1W enhance image.



**Fig. 4.** This study was performed on Giemsa banded chromosomes from phytohemagglutinin stimulated peripheral blood lymphocytes. In total 19 cells examined, all cells were normal (46, XY).

2×3 cm

가

가

(Fig. 3).

4

7

2

, Horner

7

(miosis),

(anhidrosis)

2

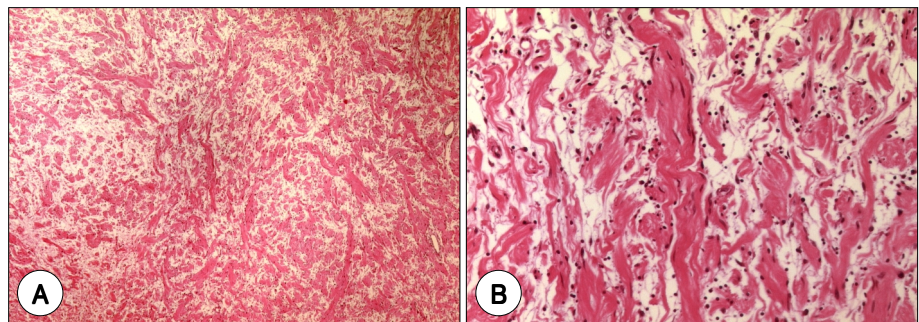
(Fig. 4) 31 10  
(neurofibroma)(Fig. 5A and B) 1 2  
(x 100) , 34  
(wavy nuclei) (Fig. 6A and B). 가  
(x 400) 가 1



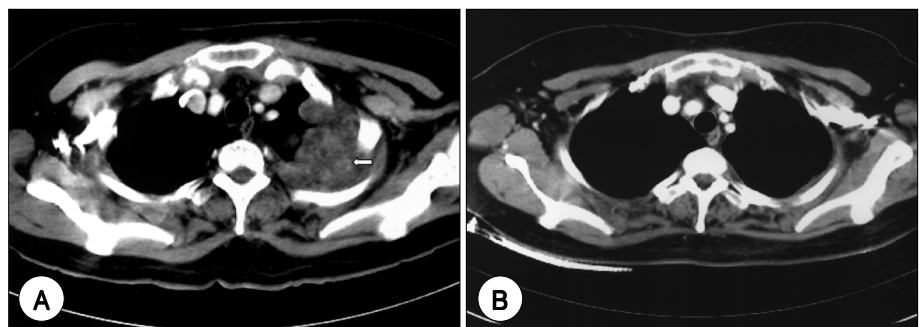
**Fig. 3.** The photograph shows multiple round shaped gray yellowish gelatinous mass. Total 31 pieces of mass were removed ; 4 in right , 20 in left deep cervical lymphatic channel area and 7 in left subclavian area. Ls : masses excised from left subclavian area.

1 1/4,000  
neurofibromin 17  
cafe - au lait ,  
(freckling),  
Lisch 2 1/100,000  
4)5) merlin( schw-  
annomin) 22  
, cafe - au lait ,  
. neurofibromin, merlin  
(tumor suppressor factor)

**Fig. 5.** A : There are markedly elongated nuclei, with a wavy, serpentine configuration and pointed ends (H-E stain, x 100). B : There are rows of cells with palisade arrangement of nuclei and intervening band of parallel wavy fibrils and stroma are edematous, like fluid collection (H-E stain, x 400).



**Fig. 6.** A : Pre-operative chest CT shows multiple conglomerated mass lesion in left lung apex, anterior and posterior chest wall. B : Post-operative chest CT shows no more mass lesion.



Korean J Otolaryngol 2004;47:1315-8

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